

# Driving toward Consensus on Optimizing Patient Care Pathway

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# Personal Perspective (And Biases!)

- Previous Chair of American College of Medical Genetics and Genomics Therapeutics Committee
  - Policy, development of evidence-based treatment guidelines
- Former Director NORD Center of Excellence, Nationwide Children's Hospital/Ohio State University, Columbus, Ohio
- Active clinical trial research in rare disorders



**CASE  
SOLVED**

**...Now what?**



# Treatment Guidance Uncertainties

- Most rare diseases do not have detailed natural history studies
- Rare disease therapeutics will continue to be challenging
  - Endpoints difficult and not always practice friendly
  - Evidence will likely always be very incomplete
- Systematic evidence-based reviews often not fruitful
  - Frequently not enough evidence for practice guideline
- Reliable source of clinical practice information still needed
- ***Patient and family experiences need to be incorporated***



# Delivering Care for Rare Disorders

- Applying clinical recommendations requires organizational structure
- Develop a process to establish a care pathway approach
  - Some disease specific CoE (CF) have good model for CoE structure
  - Many RD are very rare, not enough expert people, lack of good evidence for CoE
- Network of expertise to optimize/distribute and ensure equity of care
  - Not just specialist care - Chronic care, allied health care, etc.
  - Infrastructure – hospitals, institutes
- ***Use patient and family community to guide***



# Care Pathway Definition

1. Intervention is a structured multidisciplinary plan of care
2. Intervention is used to translate guidelines or evidence into local structures
3. Intervention details steps in a course of care (i.e., the intervention has timeframes or criteria-based progression)
4. Intervention aims to standardize care for a specific population

“the right person, in the right place, doing the right thing, at the right time, with the right outcome and all with attention to the patient experience.”

# Challenges of RD Care Pathways

- Limited abilities to recognize RD at entry to healthcare
- Lack of care pathway leads to delays (diagnosis, treatment) and inequity and places high burden of care coordination on families
- Large scope and complexity of heterogenous group of RD
- Lack of resources:
  - Financial, psychosocial support, evidence to guide care
- How to benchmark
- Integration into provincial/national systems
- Education and training

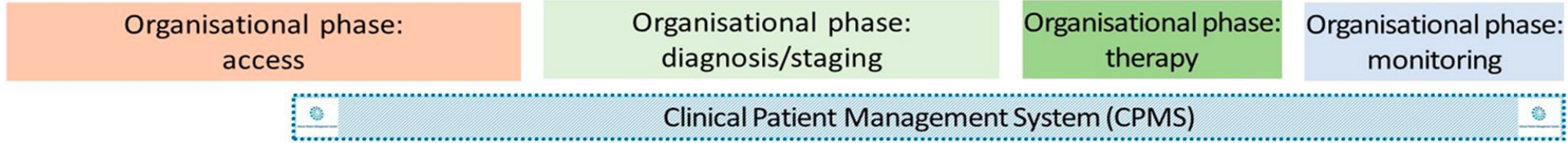
# Characteristics of RD Care Pathways

- Large load onto highly specialized services
- Centralization of services
- Balance between specialized expertise, accessibility
- Patients and families empowerment
- Blurring or boundaries between clinical care and research
- Need for complex care:
  - Psychosocial, ancillary services, community care

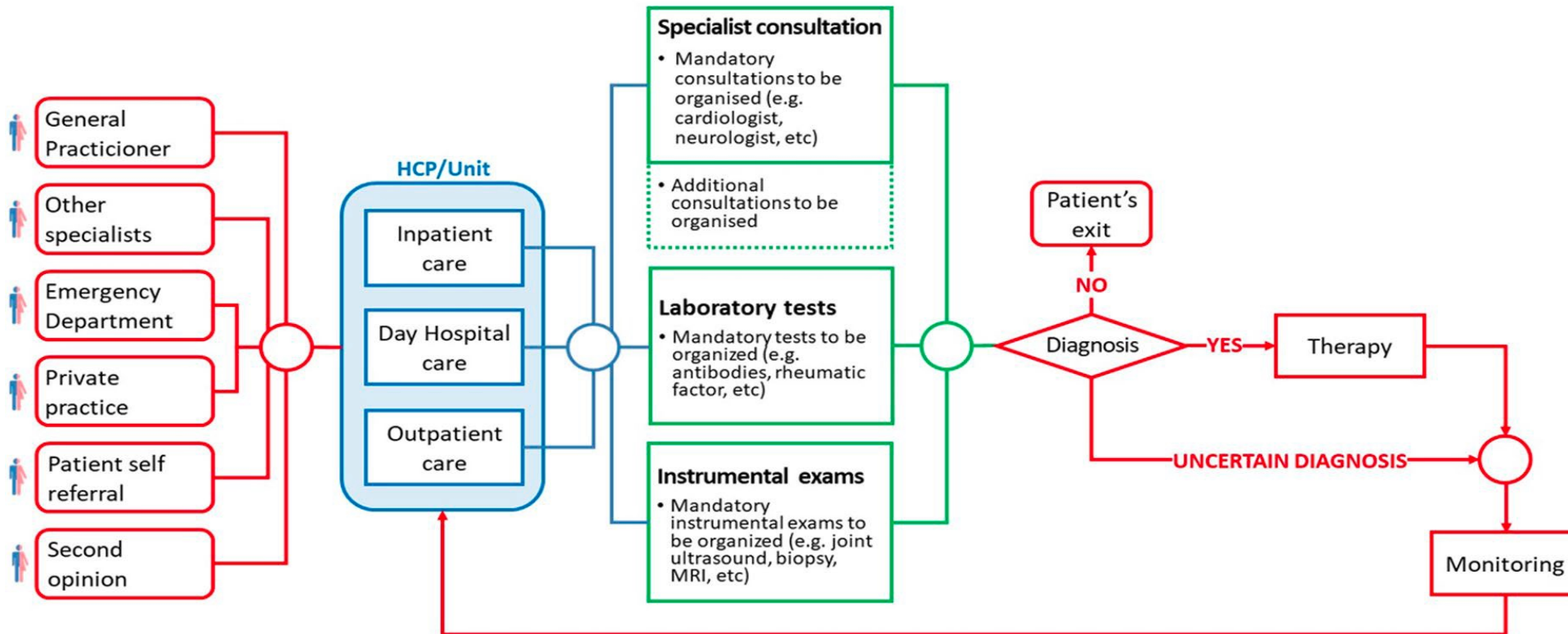




# European Reference Network on Rare and Complex Connective Tissue and Musculoskeletal Diseases

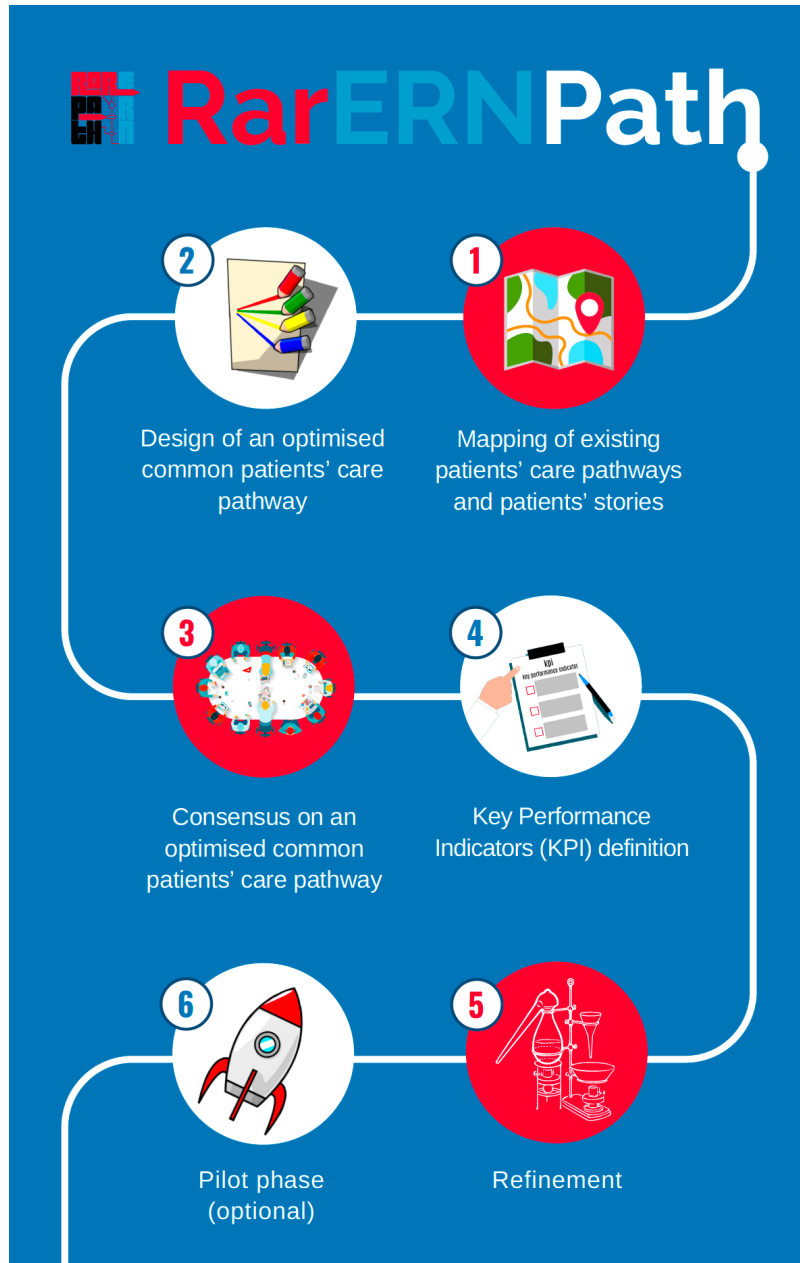


Talarico Orphanet J Rare Dis 2020





# Patient First RD Care Pathway



- European Reference Network created a method to provide care plan organization across the different EU Member States.
- Based on sharing expertise and patient-centred approach: RarERN Path™



# Concluding Thoughts on Canadian RD Care Pathway

- Patient/family centered multidisciplinary and chronic care
- Centre of Excellence model not as optimal
  - Creation of network(s) in response to need for equity of care across country and account for provincial/federal framework of health care
- Incorporation of research and education into care pathway
- Enabling access to innovative therapies